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ABSTRACT

The pamphlet uses diagrams and a question and answer format to explain briefly the causes and symptoms of Down's Syndrome and the risks of bearing children with Down's Syndrome for women over 35 years old. Discussed are such topics as typical IQ range for children with Down's Syndrome, the three genetic types of Down's Syndrome, and the function of a genetic counselor. (GW)

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facts about mongolism for women over 35

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introduction

Every pregnant woman wants to have a healthy baby. If you are planning a pregnancy and are over 35 years old, there are some facts you should know about **mongolism*** **(Down's syndrome).**

The chances of bearing a child with a **genetic defect** such as mongolism increase sharply for women over 35, and even more for those over 40.

If you are considering having children at a later time or if you have a child with mongolism in your family, this pamphlet contains answers to questions of concern to you.

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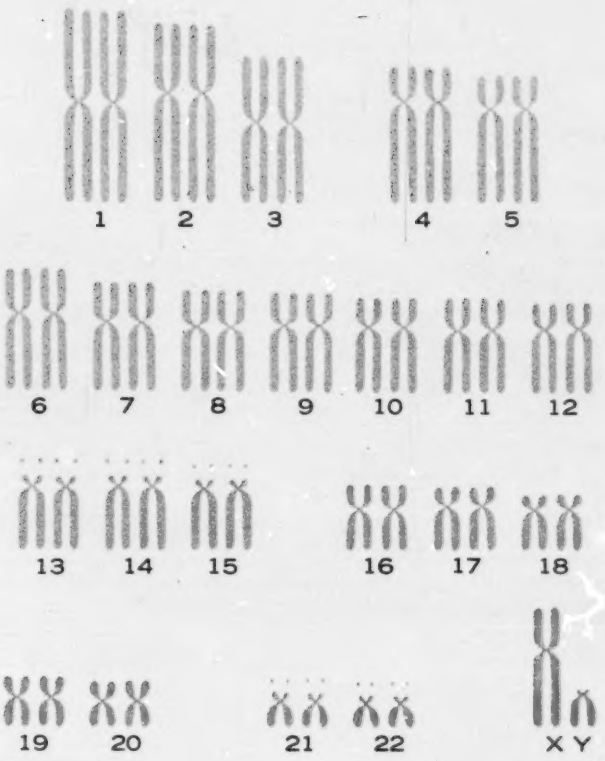
* words appearing in **bold** are explained in the Glossary

Normal Karyotype



All normal cells (except ova and sperm) have 46 chromosomes—44 autosomes and 2 sex chromosomes. Scientists have given these chromosomes numbers and have paired them according to their similarities. (Note the sex chromosome: all normal males have one "X" and one "Y" chromosome; normal females have two "X" chromosomes.)

Q. What is mongolism (Down's syndrome)?

A. Mongolism is one of the most common causes of mental retardation. It is characterized by various physical and mental abnormalities and by the presence of an extra **chromosome.**

Q. How can an infant with mongolism be identified?

A. In examining a newborn thought to have mongolism, the doctor will usually find certain telltale characteristics—slanting eyes, slightly protruding lips, small ears, larger than normal tongue, short hands, feet, and trunk, and sometimes, an unusual crease in the palm of the baby's hand. Also, babies with mongolism seem to have more heart defects as well as more frequent colds and respiratory infections than the normal infant.

Q. What is the outlook for a child with mongolism?

A. Since the advent of antibiotics and modern heart surgery, the life expectancy of a child with mongolism has greatly increased. For those with Down's syndrome who survive the first few years, death rates are about the same as for normal persons until around age 40. Because people with mongolism seem to age a little more rapidly than normal, they sometimes become susceptible to diseases associated with old age as early as age 40.

The most serious problem of children with mongolism is mental retardation. A baby with mongolism is often slow in learning to turn over, sit up, crawl, walk, and speak. In addition, he may never achieve near normal intelligence.

Society is becoming more understanding of mental retardation and has set up more educa-

tional and vocational training programs for mentally retarded individuals. At this time, there is no cure for mongolism. However, if parents can accept the initial setback of learning that their child is abnormal, they can provide good, loving homes as well as derive satisfaction from their child.

Q. How retarded will a child with mongolism be?

A. Generally, children with mongolism have IQs in the 30s and 50s, occasionally in the 60s and 70s. (Average scores for normal children are in the range of 90-110.) Studies by some scientists have shown that children with Down's syndrome who have been institutionalized have lower IQ scores than those who have remained at home. One explanation for this is that a child reared at home receives more attention and greater stimulation than one brought up in an institution. There are, however, special occasions when institutional care is in the best interests of the child and family.

Q. What causes mongolism?

A. The cause of mongolism is unknown, but it is reassuring to know that nothing a mother or father does during pregnancy can cause it.

Q. Why is mongolism called a genetic defect?

A. The child with Down's syndrome has extra genetic material, usually in the form of an entire extra chromosome. The event which results in this "extra" chromosome usually occurs during development of the **egg** or **sperm** or during **fertilization.** The extra chromosome resembles two other chromosomes designated by scientists, "pair 21." Although other genetic diseases may be associated with an extra chromosome,

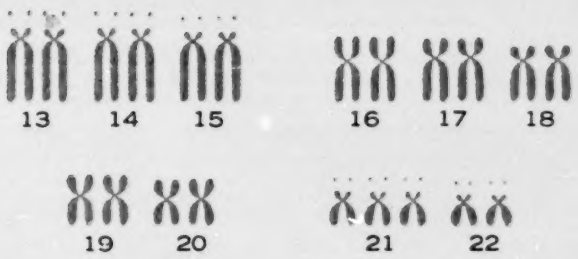
only Down's syndrome is characterized by extra "chromosome 21" material.

There are three genetic types of Down's syndrome:

1. **Trisomy 21:** This condition occurs in about 95 percent of all cases of mongolism. Though his parents have normal chromosomes, the child with trisomy mongolism has three, rather than the normal two chromosomes in pair 21. Trisomy 21 usually results from a mistake in cell division of either the sperm or egg and is an accidental occurrence. The incidence of trisomy 21 increases dramatically with the age of the mother.

2. **Translocation:** This genetic condition can take several forms, and it occurs in about four

Trisomy 21

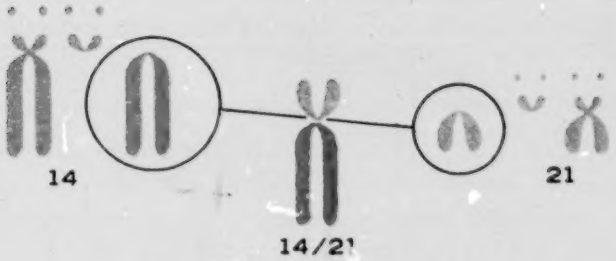


Trisomy 21 refers to a condition in which there are three rather than the usual pair of chromosomes in the "21" pair. Trisomy 21 usually results from a mistake in cell division of either the egg or the sperm and is an accidental occurrence.

percent of mongolism cases. Children with **translocation mongolism** have an extra number 21 chromosome which has broken and become attached to another chromosome. A parent can carry a translocation without showing any symptoms of disease because the parent still carries the correct amount of genetic material, although some of it is out of place (translocated).

Normal children receive only one chromosome of pair 21 from each parent. But a parent with a translocation can pass on his or her normal chromosome 21 plus the translocated chromosome 21, giving the child too much chromosome 21 material.

Formation Of A Translocation Chromosome



A translocation occurs when a piece of one chromosome breaks off and attaches to a different chromosome. This "translocation chromosome" has been formed by the breaking and rejoining of chromosomes 14 and 21 (with loss of the little pieces).

3. **Mosaicism:** This genetic type of Down's syndrome is very rare. It occurs when an individual has cells with different chromosome counts (for example, 46 in some cells and 47 in others). Mosaicism is not carried in the parents' chromosomes; it is accidental, resulting from an error which occurs in the early cell division of the fertilized egg. Babies with mosaic Down's syndrome may have only some of the features of the syndrome, because only some of their cells have an abnormal number of chromosomes.

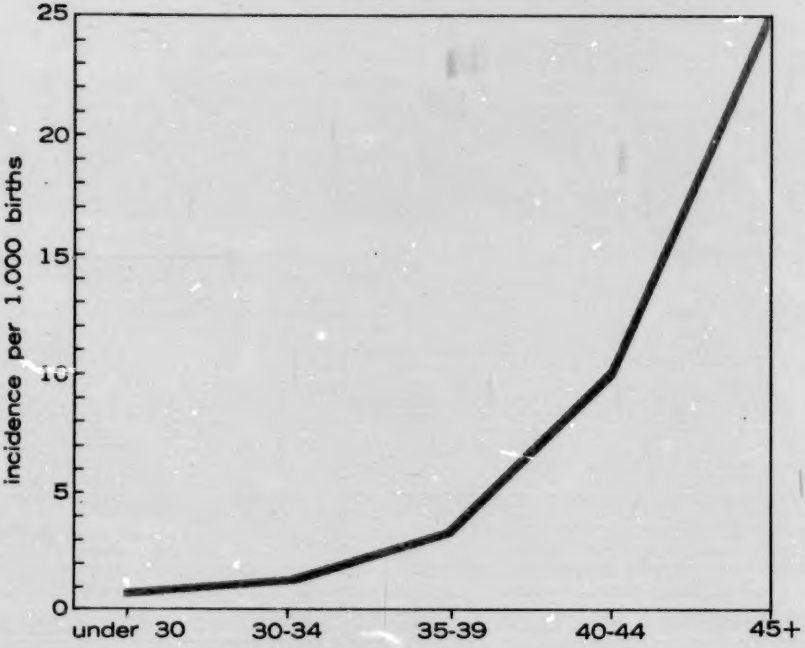
Q. What is the likelihood of having a child with mongolism?

A. The chances of bearing a baby with Down's syndrome rise with maternal age. This rise increases sharply after age 40. Although only 13 percent of all pregnancies in this country occur in women over 35, about half of the babies with Down's syndrome are born to women in this age group. The chance of bearing a baby with mongolism when you're age 30-34 is one in 750; between the ages of 35 and 39 the chance is one in 300; between the ages of 40 and 44, the chance is one in 100; and for women 45 years of age and over the chance rises to one in 40. By contrast, women under 30 years old have only one chance in 1500 of having a child with mongolism.

Q. Why does the risk of having a baby with mongolism increase with the mother's age?

A. Scientists have several theories to explain the increased risk. Women are born with all the eggs which are necessary for bearing children.

Relationship of Down's Syndrome to Maternal Age



The risk of having a child with Down's Syndrome increases with maternal age.

By the time a woman is age 35, her eggs are also 35 years old. Some scientists believe that just as people pass their prime in certain bodily functions, so may eggs.

Also, because the egg of a woman over 35 has been in the ovary for that long a time, some

scientists feel the egg is more likely to have been exposed to physical or chemical forces such as radiation which might damage the genetic material in the egg.

A third theory is based on the idea that an egg fertilized* within 24 hours after **ovulation** has the best chance of developing normally. Some older women may have intercourse less frequently and thus are a little less likely to have their eggs fertilized within the optimal 24-hour period.

Q. What can I do to have a healthy baby?

A. Take good care of your own health, even before you have children. Employ **family planning**—that is, use birth control methods so that you have only the number of children you want, when you want them. Spacing your children increases their chances of being healthy. See a physician as soon as you suspect you're pregnant and follow the diet and other guidelines he sets up for you.

Consult your family physician, gynecologist, or public health nurse IF:

- you are over age 35 OR
- you are already the parent of one child with mongolism OR
- children with genetic defects have been born in your family or your husband's family OR
- you have had great difficulty in becoming pregnant or have had several miscarriages.

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* **see fertilization** in Glossary

The doctor or health professional you consult will assess whether it is advisable for you and your husband to see a **genetic counselor.**

Q. Does a genetic counselor recommend that you don't have children?

A. No, this is a personal decision. A genetic counselor will only give you the information you need to evaluate the risk you might be facing. He may recommend that you and your husband have your chromosomes analyzed **(karyotyped).** On the basis of this analysis, he can in some cases predict the chances of your having a healthy baby. For example, if the karyotype shows that the woman has a translocation commonly associated with mongolism, the genetic counselor would be able to predict the probabilities of her child being: 1) normal; 2) normal but carrying a translocation like the mother's; or 3) a victim of Down's syndrome. The purpose of the genetic counselor is to help you understand the risk you face, so that you can plan your family thoughtfully.

If you are already pregnant, under certain circumstances your doctor or genetic counselor may suggest you undergo **amniocentesis** to find out whether the **fetus** which you are carrying has a genetic defect associated with mongolism. Amniocentesis is a technique used by specially trained physicians to obtain amniotic fluid for analysis.

GLOSSARY

Amniocentesis—A medical procedure whereby fluid is drawn from the liquid-filled sac surrounding the developing fetus while in the womb. Cells shed from the fetus into the fluid can then be specially prepared within 2-3 weeks for chromosomal analysis. (See karyotype.)

Autosomes—22 of the 23 chromosomes normally contained in ova and sperm (reproductive) cells in the human body. In all other (non-reproductive) body cells, the chromosomes occur in pairs, so each cell has 46. Each autosome usually appears identical to its partner, but each pair is different in its genetic content and frequently in its appearance from all other pairs. The remaining pair of chromosomes are the sex chromosomes and are designated XX in females and XY in males.

Chromosomes—Chromosomes are structures which are present in every human cell and are composed of genes, which carry hereditary traits. All normal cells in the human body, except ova and sperm cells, have 46 chromosomes —44 autosomes and 2 sex chromosomes. Normal reproductive cells (ova and sperm) contain 23 chromosomes. (See autosomes and sex chromosomes.)

Down's Syndrome—An alternate name for mongolism. (See mongolism.)

Egg—(See ovum.)

Family Planning—Family planning is the use of birth control methods in planning the number and timing of pregnancies.

Fertilization—Fertilization is the fusion of an egg from the woman and a sperm from the man, which then results in the start of the development of the offspring.

Fetus—The fetus is the unborn offspring. Before eleven weeks it is called an embryo. It is known as an infant or baby after it is born.

Gene—A gene is the smallest particle of hereditary information which is passed from parent to offspring. A group of genes together in a specific sequence make up the structures known as chromosomes.

Genetics—Genetics is the study of heredity or how traits (physical and mental) are passed from parents to offspring.

Genetic Counselor—A genetic counselor is an advisor who is usually qualified in several medical specialties such as internal medicine, pediatrics and genetics. The genetic counselor will compile a complete "family tree" of information about the genetic and general medical history of the patient and family. This history, together with appropriate laboratory tests, will help the genetic counselor predict the likelihood that genetic defects will occur in a family.

Genetic Defect—An abnormality of the genes. (See trisomy, translocation.)

Hereditary—Referring to traits passed from generation to generation via the genes and chromosomes.

Karyotyping—A charting system devised by scientists for numbering and pairing chromosomes

on the basis of similar characteristics such as size and shape. A karyotype is made by photographing the chromosomes, cutting out their pictures and arranging them by pairs in a standard order. Karyotyping makes chromosomes easier to study.

Mongolism—Mongolism or Down's syndrome is one of the most common causes of mental retardation. It is a combination of physical deformities and mental retardation and is characterized by a genetic defect in chromosome pair 21. (See trisomy 21, translocation mongolism, and mosaicism.)

Mosaicism—Mosaicism refers to a genetic condition in which a person's genes are different in different cells of his body. A mosaic individual can pass on normal genes or abnormal genes to the offspring.

Ovulation—Release of the egg from the ovary. (See ovum.) In most women one egg is ovulated each month.

Ovum (plural: ova)—The ova are the female egg cells which are contained in the organs known as the ovaries. Reproduction is accomplished by the successful fertilization of the ovum with a sperm and the subsequent development of the fertilized ovum into a baby. Ova (and sperm) are unlike other cells in the human body because they each have only 23 chromosomes. When they join they then can produce an offspring with the correct number, 46 chromosomes.

Sex Chromosomes—The sex chromosomes are the two chromosomes designated "X" and "Y"

by the scientists. All normal males have one "Y" and one "X" chromosome. The normal female has two "X" chromosomes and no "Y" chromosomes. The other 44 chromosomes in the normal cell are known as autosomes.

Sperm—The male reproductive cells. (See ovum.)

Translocation—The transfer of a piece of one chromosome to a different chromosome.

Translocation Mongolism—A form of mongolism which may be hereditary and which results from a piece of a chromosome in pair 21 breaking off and attaching itself to another chromosome.

Trisomy—Trisomy refers to a condition in which there are three, rather than the usual pair of chromosomes at a designated place in the karyotype.

Trisomy 21—Trisomy 21 refers to the presence of three chromosomes rather than the normal pair designated 21. This genetic defect is the chromosomal abnormality most frequently associated with mongolism. Trisomy 21 is sometimes used as an alternate name for mongolism or Down's syndrome.

* U.S. GOVERNMENT PRINTING OFFICE : 1973 O - 524-539

Much remains to be learned about genetic defects. The National Institute of Child Health and Human Development is one of many organizations concerned with genetic diseases. The Institute supports and conducts research aimed at improving each individual's growth and development and providing new and better methods of prevention, detection, and treatment of diseases, particularly those affecting mothers and children.

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